5. **Bickerstaff Brainstem Encephalitis (BBE)**
    - Overlaps with MFS but includes central nervous system involvement, particularly the brainstem.
    - Symptoms include altered consciousness and long tract signs.
    - Paralysis: Similar to MFS with additional central nervous system symptoms.
6. **Pharyngeal-Cervical-Brachial Variant**
    - Involves weakness in the pharynx, neck, and arms.
    - Often presents with difficulty swallowing and speaking.
    - Paralysis: Affects the muscles in the pharyngeal, cervical, and brachial areas.
7. **Acute Panautonomic Neuropathy**
    - Rare form affecting the autonomic nervous system.
    - Symptoms include cardiovascular irregularities, gastrointestinal disturbances, and urinary retention.
    - Paralysis: Not typically characterized by motor paralysis but by autonomic dysfunction.
8. **Sensory Guillain-Barré Syndrome**
    - Rare variant where sensory nerves are predominantly affected.
    - Paralysis: Motor function is largely preserved, but significant sensory deficits are present.

Common Features Across Variants

- ***Progression***: Typically, the peak of weakness is reached within two to four weeks.
- ***Recovery***: Recovery can take weeks to years, and while many patients fully recover, some may have long-term deficits.
- ***Treatment***: Intravenous immunoglobulins (IVIG) or plasmapheresis are common treatments across all types.

The type and pattern of paralysis in Guillain-Barré Syndrome vary depending on the specific subtype.

Subtype	Type of Paralysis	Key Features
Acute Inflammatory Demyelinating Polyneuropathy (AIDP)	Ascending paralysis (starts in legs)	Symmetrical muscle weakness Sensory loss and tingling sensations
Acute Motor Axonal Neuropathy (AMAN)	Motor paralysis without sensory involvement	Rapid muscle weakness Affects primarily motor neurons
Acute Motor-Sensory Axonal Neuropathy (AMSAN)	Motor and sensory paralysis	Severe form Affects both motor and sensory nerves Slow recovery
Miller Fisher Syndrome (MFS)	Ophthalmoplegia (eye muscle paralysis)	Ataxia (lack of muscle coordination) Areflexia (absence of reflexes)
Bickerstaff Brainstem Encephalitis (BBE)	Similar to MFS with central nervous system symptoms	Altered consciousness Long tract signs
Pharyngeal-Cervical-Brachial Variant	Affects pharynx, neck, and arms	Difficulty swallowing and speaking
Acute Panautonomic Neuropathy	Autonomic dysfunction, not motor paralysis	Cardiovascular irregularities Gastrointestinal disturbances Urinary retention
Sensory Guillain-Barré Syndrome	Sensory deficits, motor function preserved	Significant sensory deficits without major motor paralysis

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Q: Management of a child with Herpex simplex virus (HSV) encephalitis

- ❖ HSV encephalitis in children is a medical emergency requiring prompt diagnosis and aggressive treatment.
- ❖ The mainstay of therapy is intravenous acyclovir, along with comprehensive supportive care and monitoring for complications.
- ❖ Early intervention and a multidisciplinary approach are crucial for improving outcomes and minimizing long-term sequelae.

Initial Assessment and Stabilization

- ***Clinical Evaluation:*** Assess for signs of increased intracranial pressure, seizures, altered mental status, and focal neurological deficits.
- ***Laboratory Tests:*** Include complete blood count, electrolytes, liver function tests, and coagulation profile.

- ***EEG in case of seizures –*** temporal lobe focality
- ***Lumbar Puncture:*** For cerebrospinal fluid (CSF) analysis, including PCR for HSV DNA, which is the diagnostic gold standard.
- ***Imaging:*** MRI of the brain with and without contrast is preferred over CT to identify characteristic temporal lobe changes.

Antiviral Therapy

- ***Acyclovir:*** The first-line treatment.
    - ***Dosage:*** High-dose intravenous acyclovir (60 mg/kg/day divided every 8 hours) for a minimum of 14-21 days.
    - ***Monitoring:*** Renal function should be monitored due to the risk of nephrotoxicity.
- ***Consideration of Other Antivirals:*** In cases of acyclovir resistance (rare in immunocompetent children), alternatives like foscarnet or cidofovir can be considered.

Supportive Care

- ***Management of Increased Intracranial Pressure:*** May include head elevation, sedation, and mannitol or hypertonic saline.
- ***Seizure Control:*** Antiepileptic drugs for seizure management or prophylaxis if there is a high risk of seizures. Commonly phenytoin and CBZ, oxcarb are used
- ***Temperature and Blood Pressure Management:*** Fever should be controlled; blood pressure should be monitored and managed appropriately.
- ***Fluid and Electrolyte Balance:*** Careful monitoring and management to avoid complications like cerebral edema or dehydration.
- ***Nutritional Support:*** Enteral feeding if the child is unable to eat.

Monitoring for Complications

- ***Neurological Monitoring:*** Regular neurological assessments to detect changes in the level of consciousness, new onset of seizures, or focal deficits.
- ***Long-term Neurological Sequelae:*** Includes cognitive impairment, motor deficits, and epilepsy. Early rehabilitation and regular follow-up are essential.

Follow-up and Rehabilitation

- ***Neurological Follow-up:*** Regular assessments to monitor recovery and detect any long-term sequelae.

- ***Rehabilitation Services:*** May include physical therapy, occupational therapy, and speech therapy, depending on the deficits.
- ***Psychological Support:*** For the child and family, considering the potential impact on mental health.

Prevention and Prophylaxis

- ***Prophylactic Antiviral Therapy:*** In neonates with HSV exposure or in cases of recurrent HSV encephalitis.
- ***Education of Caregivers:*** About the nature of the disease and the importance of adherence to therapy and follow-up appointments.

Research and Emerging Therapies

- Ongoing research into new antiviral agents and vaccine development.
- Investigational use of immunomodulatory therapies like steroids and intravenous immunoglobulin (IVIG).

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Q: Guillain-Barré Syndrome (GBS)

- ❖ Guillain-Barre Syndrome (GBS) is an acute polyradiculoneuropathy, often post-infectious, leading to varying degrees of weakness, sensory abnormalities, and autonomic dysfunction
- ❖ GBS in children is a rare but serious condition requiring prompt recognition and treatment
- ❖ The mainstay of management is supportive care and immunotherapy
- ❖ Most children recover, but the disease can be life-threatening, especially if respiratory muscles are involved
- ❖ Long-term follow-up is essential to address any residual deficits and provide necessary rehabilitation and support.

Epidemiology

- ***Incidence:*** Relatively rare in children compared to adults.
- ***Age Distribution:*** Can occur at any age, but less common in very young children.
- ***Gender Predilection:*** Slightly more common in males.

Etiology

- ***Post-Infectious:*** Often follows respiratory or gastrointestinal infections.

- ***Associated Pathogens:*** Campylobacter jejuni, Epstein-Barr virus, Cytomegalovirus, and Mycoplasma pneumoniae are common precedents.
- ***Immunization:*** Rarely associated with immunizations.

Pathophysiology

- ***Immune-Mediated:*** Autoimmune attack on peripheral nerves and their myelin sheaths, sometimes on axons.
- ***Molecular Mimicry:*** The immune system attacks nerves mistaking them for infectious agents.
- ***Inflammatory Response:*** Leads to demyelination or axonal damage, impairing nerve signal transmission.

Clinical Manifestations

- ***Onset:*** Acute, often 1-3 weeks post-infection.
- ***Symptoms Progression:*** Ascending weakness, starting in the legs.
- ***Muscle Weakness:*** Symmetrical; can progress to paralysis.
- ***Sensory Symptoms:*** Tingling, numbness; usually less prominent than motor symptoms.
- ***Autonomic Dysfunction:*** Can include blood pressure fluctuations, cardiac arrhythmias, and urinary retention.
- ***Respiratory Involvement:*** May require prolonged mechanical ventilation in severe cases.
- ***Pain:*** Common, often neuropathic.

Diagnosis

- ***Clinical Assessment:*** Based on history and neurological examination.
- ***Lumbar Puncture:*** Elevated protein in cerebrospinal fluid (CSF) with normal cell count (albuminocytologic dissociation).
- ***Electrophysiological Studies:*** Show demyelinating or axonal neuropathy.
- ***Exclusion of Other Causes:*** Important to rule out mimics like transverse myelitis or acute myelopathy.

Treatment

- ***Supportive Care:*** Respiratory support if needed, pain management, and prevention of complications like deep vein thrombosis.
- ***Immunotherapy:***
    - ***Intravenous Immunoglobulin (IVIG):*** Often first-line; reduces severity and duration.
    - ***Plasmapheresis (Plasma Exchange):*** Alternative to IVIG, especially if started early.
- ***Steroids:*** Not typically recommended for GBS.
- ***Rehabilitation:*** Physical and occupational therapy for recovery.

Prognosis

- ***Recovery:*** Most children have a good prognosis with recovery starting within weeks to months.
- ***Residual Weakness:*** Some may have lingering deficits.
- ***Relapse:*** Rare in children.
- ***Mortality:*** Low in children, but higher in severe cases with respiratory involvement.

Complications

- ***Respiratory Failure:*** Due to diaphragmatic and intercostal muscle weakness.
- ***Autonomic Dysfunction:*** Can lead to dangerous fluctuations in blood pressure and heart rate.
- ***Pain:*** Can be severe and require management.
- ***Deep Vein Thrombosis (DVT):*** Due to immobility.
- ***Pressure Ulcers:*** From prolonged bed rest.

Long-term Management

- ***Monitoring:*** For signs of relapse or chronic neuropathy.
- ***Psychological Support:*** For the child and family, addressing the impact of the disease and prolonged recovery period.

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Q: Indications of neuroimaging in a child with headache.

- ❖ The decision to perform neuroimaging in a child with headache should be based on a thorough clinical evaluation.
- ❖ Not all headaches require imaging, but certain features and associated symptoms increase the likelihood of an underlying pathological cause that warrants further investigation.
- ❖ The choice between MRI and CT should be guided by the clinical scenario, availability, and urgency of the situation.
- ❖ Early and appropriate imaging can significantly impact the management and outcome in cases where structural or serious intracranial pathology is present.

General Considerations

- ***Age:*** Younger children, especially under 3 years, are more likely to require imaging.
- ***Headache Characteristics:*** Sudden onset, increasing frequency or severity, or a headache that is different from their usual pattern.
- ***Associated Symptoms:*** Nausea, vomiting, visual disturbances, or other neurological deficits.
- ***Family History:*** Consider imaging if there is a family history of neurologic diseases, bleeding tendancy or tumors.

Specific Indications for Neuroimaging

1. ***Abnormal Neurological Examination:***
    - Focal neurological deficits.
    - Signs of increased intracranial pressure (ICP), such as papilledema.
    - Altered mental status.
    - Cranial nerve abnormalities.
2. ***Headache Type and Pattern:***
    - Thunderclap headache (sudden, severe headache).
    - New, persistent headache in the morning.
    - Headache waking the child from sleep.
    - Headache exacerbated by Valsalva maneuvers, coughing, or exercise.
3. ***Change in Headache Characteristics:***

- Significant change in the frequency, intensity, or character of the headache.
- Headache that progressively worsens over weeks or months.

4. ***Associated Conditions:***
   - Presence of seizures.
   - Symptoms suggestive of a systemic illness (e.g., fever, weight loss).
   - Neurocutaneous markers (e.g., café-au-lait spots, indicative of neurofibromatosis).
5. ***History of Head Trauma:***
   - Especially if significant or with loss of consciousness.
6. ***Presence of Risk Factors:***
   - History of cancer or immunodeficiency.
   - Family history of hereditary syndromes associated with brain tumors.
7. ***Atypical Headache Syndromes:***
   - Occipital headaches in children (rare and more likely to be associated with structural pathology).
   - Chronic daily headaches not responding to treatment.

Imaging Modality Choice

- ***MRI:*** Preferred for its detailed visualization of brain structures, vasculature, and absence of radiation. Especially useful in evaluating posterior fossa, pituitary gland, and cranial nerves.
- ***CT Scan:*** Used when MRI is not available or in emergency situations (e.g., suspected hemorrhage, acute trauma). Faster but involves radiation.

Red Flags Warranting Urgent Imaging

- Sudden onset of severe headache.
- Headache with signs of increased ICP.
- Neurological deficits accompanying the headache.
- Headache following a head injury.

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Q: Causes of hydrocephalus according to the level of obstruction in CSF flow

- Hydrocephalus, characterized by an abnormal accumulation of cerebrospinal fluid (CSF) within the ventricles of the brain, can be caused by various factors depending on the level of obstruction in the CSF flow
- The causes of hydrocephalus are diverse and depend on the anatomical level of obstruction or dysfunction in the CSF pathway
- Understanding the underlying cause is crucial for appropriate management, which may include surgical intervention like ventriculoperitoneal shunt placement or endoscopic third ventriculostomy, and addressing the primary pathology (e.g., tumor removal, treatment of infection)
- Early diagnosis and treatment are essential to prevent or minimize potential neurological damage and improve outcomes.

1. Obstruction at the Level of Lateral and Third Ventricles

- ***Intraventricular Hemorrhage:*** Common in premature infants, leading to post-hemorrhagic hydrocephalus.
- ***Tumors:*** Such as colloid cysts, craniopharyngiomas, or gliomas obstructing the foramen of Monro or third ventricle.
- ***Congenital Stenosis:*** Narrowing of the foramina of Monro.
- ***Infections:*** E.g., ventriculitis, which can cause scarring and narrowing.

2. Obstruction at the Level of Aqueduct of Sylvius (Aqueductal Stenosis)

- ***Congenital Aqueductal Stenosis:*** A common cause of congenital hydrocephalus.
- ***Tumors:*** E.g., tectal gliomas.
- ***Inflammation/Infection:*** Leading to scarring and narrowing of the aqueduct.
- ***Hemorrhage:*** In the tectal region.

3. Obstruction at the Level of Fourth Ventricle

- ***Tumors:*** Medulloblastomas, ependymomas, or brainstem gliomas.
- ***Chiari Malformations:*** Particularly Chiari II malformation.
- ***Arachnoid Cysts:*** Can compress the fourth ventricle.
- ***Infections and Hemorrhage:*** Leading to scarring and obstruction at the outlet foramina (Luschka and Magendie).

4. Obstruction in the Subarachnoid Space (Communicating Hydrocephalus)

- ***Meningitis:*** Bacterial, viral, or tubercular meningitis causing scarring and impaired CSF absorption.
- ***Subarachnoid Hemorrhage:*** Leading to fibrosis in the subarachnoid space.
- ***Post-Surgical Scarring:*** Following brain or spinal surgery.
- ***Arachnoid Villi Blockage:*** Due to various causes like tumors or cysts.

5. Overproduction of CSF (Rare)

- ***Choroid Plexus Papilloma:*** A tumor that can produce excessive CSF.

6. Impaired CSF Absorption

- ***Venous Hypertension:*** Impairing CSF absorption in the arachnoid villi.
- ***Idiopathic Intracranial Hypertension:*** Previously known as pseudotumor cerebri.

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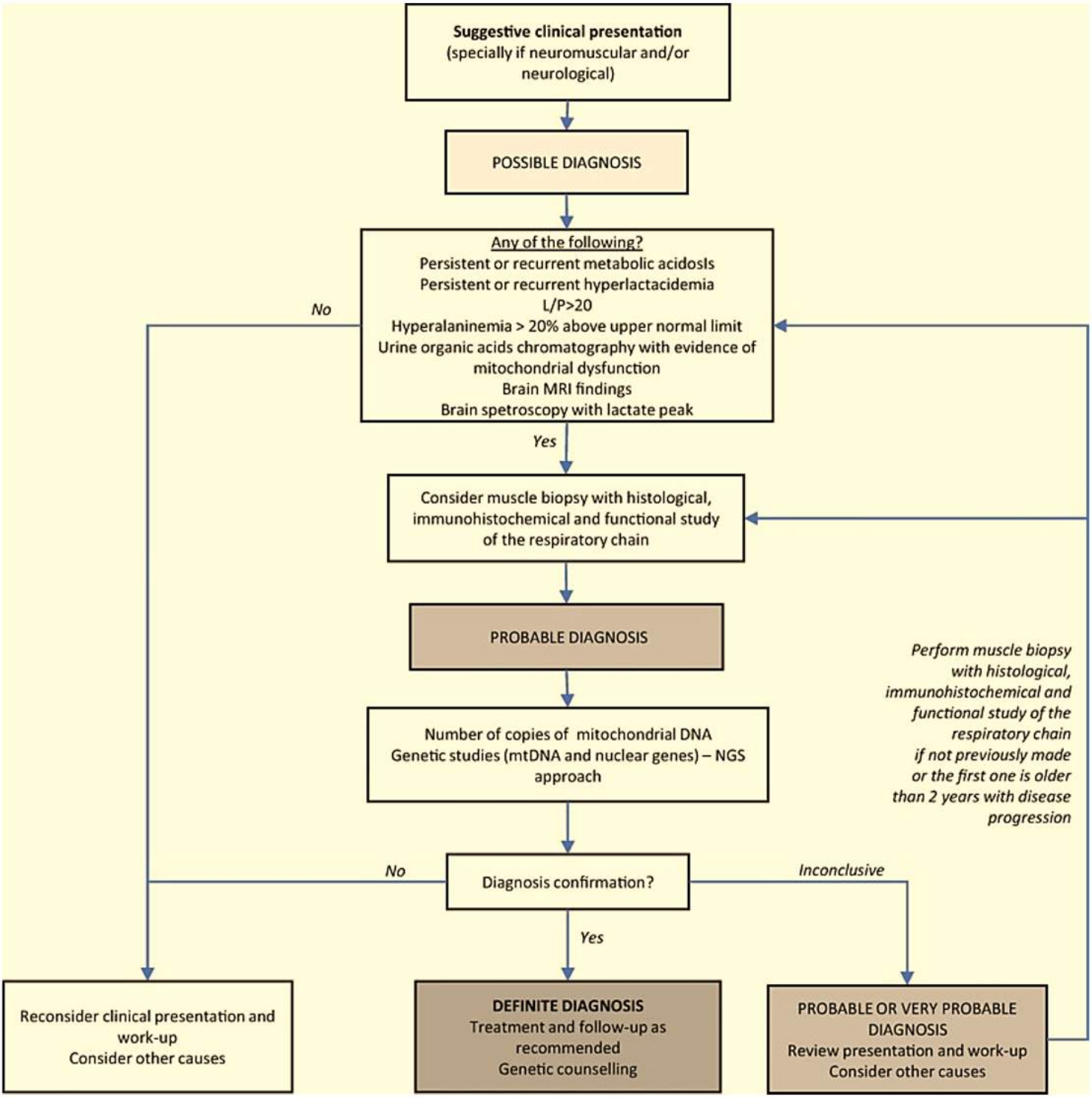
Q: Diagnostic algorithm for suspected mitochondrial disease

- ***Clinical Presentation***: The algorithm begins with the recognition of suggestive clinical signs, particularly neuromuscular and/or neurological symptoms which are characteristic of mitochondrial disorders.
- ***Possible Diagnosis***: If such clinical presentation is observed, the algorithm considers a possible diagnosis of mitochondrial disease.
- ***Initial Investigations***: It then guides the clinician to look for specific biochemical markers such as:
    - Persistent or recurrent metabolic acidosis
    - Persistent or recurrent hyperlactatemia (>2 mmol/L)
    - Hyperalaninemia (>20% above the upper normal limit)
    - Organic acid chromatography showing abnormalities
    - Brain MRI findings indicative of mitochondrial dysfunction
    - Brain spectroscopy with a lactate peak
- ***Further Evaluation***: If any of these findings are present, the algorithm advises to consider a muscle biopsy for histological, immunohistochemical, and functional study of the respiratory chain, which are more definitive tests.

- ***Probable Diagnosis***: If the findings from the muscle biopsy support mitochondrial dysfunction, a probable diagnosis is made.
- ***Definitive Genetic Testing***: The next step involves genetic testing to quantify the number of copies of mitochondrial DNA and to conduct comprehensive studies of both mitochondrial and nuclear genes using next-generation sequencing (NGS) techniques.
- ***Diagnosis Confirmation***: If the genetic testing confirms the diagnosis, a definitive diagnosis of mitochondrial disease is made.
- ***Treatment and Follow-up***: Following a definitive diagnosis, appropriate treatment is initiated, and follow-up is recommended, along with genetic counseling due to the inheritable nature of many mitochondrial disorders.
- ***Inconclusive Results***: If the muscle biopsy or genetic testing results are inconclusive, the algorithm suggests a review of the presentation and work-up, and consideration of other causes.
- ***Reevaluation***: If the diagnosis is not confirmed and remains uncertain after initial testing, clinicians are advised to reconsider the clinical presentation and work-up and to consider alternative diagnoses.


Suggestive clinical presentation
(specially if neuromuscular and/or neurological)
POSSIBLE DIAGNOSIS
Any of the following?
Persistent or recurrent metabolic acidosIs
Persistent or recurrent hyperlactacidemia
L/P>20
Hyperalaninemia > 20% above upper normal limit
Urine organic acids chromatography with evidence of mitochondrial dysfunction
Brain MRI findings
Brain spetroscopy with lactate peak
No
Yes
Consider muscle biopsy with histological, immunohistochemical and functional study of the respiratory chain
PROBABLE DIAGNOSIS
Perform muscle biopsy with histological, immunohistochemical and functional study of the respiratory chain if not previously made or the first one is older than 2 years with disease progression
Number of copies of mitochondrial DNA
Genetic studies (mtDNA and nuclear genes) – NGS approach
No
Diagnosis confirmation?
Inconclusive
Yes
Reconsider clinical presentation and work-up
Consider other causes
DEFINITE DIAGNOSIS
Treatment and follow-up as recommended
Genetic counselling
PROBABLE OR VERY PROBABLE DIAGNOSIS
Review presentation and work-up
Consider other causes

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Q: Early diagnosis and intervention in cerebral palsy

- Early diagnosis and intervention in cerebral palsy (CP) are crucial for optimizing outcomes and improving the quality of life for affected children
- Early diagnosis and intervention in CP are multidisciplinary efforts aimed at maximizing the child's potential
- They involve a combination of therapeutic, educational, and supportive strategies, tailored to the individual needs of the child and their family
- Regular follow-up and adaptation of interventions as the child grows are essential for optimal outcomes.

Early Diagnosis of Cerebral Palsy

1. ***Risk Factor Identification***:
    - Prematurity, low birth weight, birth asphyxia, intrauterine infections, and neonatal strokes are significant risk factors.
    - Prenatal history and perinatal events should be reviewed.
2. ***Developmental Surveillance***:
    - Regular monitoring of developmental milestones.
    - Early identification of motor delays or asymmetries.
3. ***Neurological Examination***:
    - Assessment of muscle tone, reflexes, motor control, and posture.
    - Observation of abnormal movements or asymmetries.
4. ***Use of Screening Tools***:
    - Tools like the General Movements Assessment in infants can predict CP.
5. ***Parental Concerns***:
    - Parents often first to notice delays or abnormalities in movement or development.
6. ***Imaging Studies***:
    - MRI of the brain to identify structural abnormalities or brain injuries.
7. ***Multidisciplinary Evaluation***:
    - Involvement of pediatric neurologists, physiotherapists, and developmental pediatricians.

Early Intervention in Cerebral Palsy

1. ***Physical Therapy***:
    - Focus on improving motor function, strength, and flexibility.
    - Techniques include stretching, strengthening exercises, and motor skill training.
2. ***Occupational Therapy***:
    - Aims to enhance daily living skills, including self-care and fine motor skills.
    - Use of adaptive equipment and techniques to promote independence.
3. ***Speech and Language Therapy***:
    - For those with speech and swallowing difficulties.
    - Includes communication aids for non-verbal children.
4. ***Early Educational Interventions***:
    - Special education programs tailored to individual needs.
    - Focus on cognitive, social, and emotional development.
5. ***Orthopedic Management***:
    - Management of musculoskeletal complications like contractures and deformities.
    - May include orthotics, splints, or surgery.
6. ***Medication***:
    - For spasticity management: Baclofen, Botulinum toxin injections.
    - Management of associated conditions like epilepsy.
7. ***Family Support and Counseling***:
    - Education about CP and its management.
    - Psychological support for families.
8. ***Regular Monitoring and Assessment***:
    - Ongoing assessment of developmental progress and functional abilities.
    - Adjustments in intervention plans based on child's evolving needs.

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Q: Ketogenic diet in management of Refractory seizures

- The ketogenic diet is a specialized, high-fat, low-carbohydrate diet that has been used for the management of refractory seizures, particularly in children
- The ketogenic diet is a valuable treatment option for managing refractory seizures in children, with potential benefits extending beyond seizure control
- Its implementation requires careful medical supervision, dietary planning, and monitoring for side effects
- While not without challenges, for many patients, the ketogenic diet can significantly improve quality of life where other treatments have failed.

Basics of the Ketogenic Diet

1. ***Composition***:
    - High fat, adequate protein, and very low carbohydrate.
    - Typically, the ratio of fats to carbohydrates and proteins is 4:1 or 3:1.
2. ***Mechanism of Action***:
    - Mimics the state of fasting, leading the body to burn fats rather than carbohydrates.
    - Produces ketone bodies which are thought to have an anticonvulsant effect.
3. ***Types of Ketogenic Diets***:
    - Classic Ketogenic Diet: Strict, with precise measurement of food intake.
    - Modified Atkins Diet: Less restrictive, easier to follow, lower fat-to-carb ratio.
    - MCT (Medium Chain Triglyceride) Diet: Uses MCT oil to induce ketosis, allowing more carbs and protein.

Indications

1. ***Refractory Epilepsy***:
    - Children who have not responded to two or more antiepileptic drugs.
    - Particularly effective in certain syndromes like Dravet Syndrome, Lennox-Gastaut Syndrome.

2. ***Other Neurological Disorders***:
    - Some evidence for effectiveness in GLUT1 deficiency, tuberous sclerosis complex, and Rett syndrome.

Implementation

1. ***Medical Supervision***:
    - Initiation and maintenance under strict medical supervision.
    - Regular monitoring of growth, nutritional status, and side effects.
2. ***Hospital Admission***:
    - Often started in a hospital setting to monitor for initial side effects and efficacy.
3. ***Dietary Planning***:
    - Tailored by a dietitian to ensure nutritional needs are met.
    - Gradual introduction of the diet over several days.
4. ***Education and Support***:
    - Training for caregivers in diet management.
    - Ongoing support from a dietitian and medical team.

Efficacy

1. ***Seizure Reduction***:
    - Many children experience a significant reduction in seizure frequency.
    - Some become seizure-free, though this is less common.
2. ***Cognitive and Behavioral Improvements***:
    - Improvements in alertness, behavior, and cognitive functioning reported in some cases.

Side Effects and Management

1. ***Gastrointestinal Issues***:
    - Constipation, gastroesophageal reflux, and vomiting.
    - Managed with dietary adjustments and medications.
2. ***Nutritional Deficiencies***:
    - Risk of deficiencies in vitamins and minerals.

- Supplementation often necessary.

3. ***Bone Health***:
    - Potential impact on bone density.
    - Monitoring and supplementation may be required.
4. ***Lipid Profile Changes***:
    - Can cause elevated cholesterol and triglycerides.
    - Regular monitoring of lipid profile is recommended.
5. ***Nephrolithiasis***:
    - Increased risk of kidney stones.
    - Adequate hydration and specific medications can reduce risk.

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Q: Mitochondrial disorders

- Mitochondrial disorders are a group of conditions caused by dysfunction in the mitochondria and can be challenging to diagnose and manage.
- They often present with elevated lactate levels due to impaired oxidative phosphorylation.
- Management of mitochondrial disorders is primarily supportive and symptomatic, as there are no definitive cures.
- It often requires a multidisciplinary approach involving neurologists, geneticists, cardiologists, ophthalmologists, and other specialists.
- Genetic counseling is important for affected families due to the hereditary nature of these disorders.
- Research is ongoing into novel therapies, including gene therapy and mitochondrial replacement techniques.
- Regular follow-up and monitoring of disease progression are crucial, and treatment plans may need to be adjusted over time.

Mitochondrial Disorder	Clinical Features	Diagnostic Tools	Management Strategies
MELAS (Mitochondrial Encephalopathy, Lactic Acidosis, and Stroke-like episodes)	Stroke-like episodes, seizures, muscle weakness, migraines, hearing loss	Muscle biopsy, genetic testing (often MT-TL1 gene mutation)	Symptomatic treatment (anticonvulsants, hearing aids), Coenzyme Q10, L-arginine during stroke-like

			episodes, avoidance of metabolic stressors
MERRF (Myoclonic Epilepsy with Ragged Red Fibers)	Myoclonus, epilepsy, ataxia, muscle weakness, hearing loss	Muscle biopsy (ragged red fibers), genetic testing (commonly MT-TK gene mutation)	Symptomatic treatment (antiepileptics, physical therapy), Coenzyme Q10, L-carnitine, vitamins
Leigh Syndrome	Psychomotor retardation, hypotonia, respiratory issues, ataxia, ophthalmoplegia	MRI (showing lesions in basal ganglia and brainstem), genetic testing (various nuclear and mitochondrial genes)	Supportive care (respiratory support, nutritional support), vitamins (thiamine, riboflavin), Coenzyme Q10
Kearns-Sayre Syndrome	Ptosis, ophthalmoplegia, retinopathy, heart block, cerebellar ataxia	Genetic testing, ECG, eye examination	Supportive care (pacemaker for heart block, eyelid surgery), vitamins and supplements (Coenzyme Q10, vitamins C and E)
NARP (Neuropathy, Ataxia, and Retinitis Pigmentosa)	Neuropathy, ataxia, retinitis pigmentosa, seizures	Genetic testing (often MT-ATP6 gene mutation), electroretinogram	Symptomatic treatment (antiepileptics, visual aids), physical therapy, vitamins and supplements
LHON (Leber's Hereditary Optic Neuropathy)	Bilateral, painless, subacute visual loss	Genetic testing (common mutations in MT-ND1, MT-ND4, MT-ND6 genes), ophthalmologic evaluation	Idebenone (antioxidant therapy), supportive visual aids, gene therapy (experimental)
Alpers-Huttenlocher Syndrome	Progressive neurodegeneration, seizures, liver dysfunction	Liver biopsy, genetic testing (POLG gene mutations)	Symptomatic treatment (antiepileptics, liver support), avoidance of valproate and other hepatotoxic drugs